sions which spontaneously granulated in. Thus they did not require operation and still survived. However, nearly all children not operated upon will die. If the primary physician, or other personnel involved, advises against repair of the lesions, then there must be some method of euthanasia so that such children do not constitute a burden to either the parents or the state.

After the spinal lesion has been closed and the problems of meningitis are gone, the neurosurgical problem that then remains is control of the hydrocephalus which usually develops. The onset of hydrocephalus in the newborn is apparent either at birth or within the first month or so. This is important for the primary physician to realize. The head should be measured, the fontanelle should be inspected and palpated to determine tenseness, and the findings should be recorded so that a week or two weeks later it can be determined whether there has been a change. Many times a baby is seen at one or two months of age and there is a question as to whether hydrocephalus exists or not, but there is no previous head circumference measurement available for comparison, even though the ventricles are in fact beginning to dilate. As far as the remainder of the neural tube is concerned, there may be associated defects anywhere which can cause hydrocephalus; they can be at the aqueduct, as is often the case in the Arnold-Chiari malformation, or in the posterior fossa. Occasionally, failure of development of the proper system of arachnoidal villi to return spinal fluid to the blood circulation is postulated. A block anywhere in the circulation of cerebrospinal fluid will produce hydrocephalus, and anything which obstructs the one-way flow of spinal fluid as it enters the venous system does the same thing although more slowly.

There are many terms, such as communicating hydrocephalus (so called because a dye placed in the ventricular system can be recovered in the spinal fluid), but that does not necessarily mean that there is no obstruction around the base of the system, such as in a postmeningitis situation. Any block, depending on its location, may cause enlargement of the head from increased intracranial pressure. It is obvious that a block of the aqueduct will cause a very rapid ballooning of the ventricles. A block around the base of the system may be partial, so that head growth may be only very slightly increased over normal, and it may take a long time to make the diagnosis that the ventricles are abnormally enlarged.

The present method of treatment is to place a shunt that goes from the ventricular system either to the peritoneum or to the blood stream through the jugular vein, reaching the auricle via the superior vena cava. Several types of shunts are available, some with a palpable reservoir, placed in a bur hole, that provides a little pumping system. When a shunt is made in a child with spina bifida, the meningocele itself in many instances will collapse and make it easier to treat, but there is a converse problem in that if infection is present, it sometimes will get into the system and thereby cause the very complication that it is intended to avoid, namely, infection of the shunt itself, with subsequent obstruction to spinal fluid flow. The decision regarding therapy is a complex problem; each case must be dealt with by first making an accurate evaluation of flow and then trying to see which approach appears best. In many instances a shunt is not the best answer for a child with minimal hydrocephalus. There are many reasons for this, relating of course to the possibility of postoperative infection and also to the effects on the ventricular wall in a marginal hydrocephalus that may already be arrested. Problems often occur in maintaining proper function of shunts, but if we can control the hydrocephalus after the lesion has been repaired, then the real threat to life is urinary tract infection.

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## MYELOMENINGOCELE—PART VI

# General Considerations

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SPINA BIFIDA is a disease entity which includes a group of developmental defects of the spinal column in which there is failure of fusion of vertebral arches, with or without protrusion and dysplasia

Part VI of an article in six parts.

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of the spinal cord and its membranes. The term *rachischisis* has been used to refer to this condition, since it indicates a congenital fissure of the spinal column (from the Greek *rachis,* spine, and *schisis,* cleft). A variety of defects can occur in spina bifida:

*Spina bifida occulta,* in which there is a lack of fusion of vertebral arches without any cystic distension of the meninges, is a common finding in normal children, particularly frequent being fusion defects of the axis and the fifth lumbar, first sacral, and last sacral segments. It may be quite extensive and yet produce no symptoms or signs. The skin over the defect is generally normal but may show a pigmented nevus, a hairy patch, a small lipoma, a dimple, a hemangioma, a scar or, occasionally, a palpable bony defect.

*Spina bifida cystica* (SBC) may be divided into two categories—(a) meningocele, in which there is lack of fusion of the vertebral arches, with cystic distension of the meninges but absence of myelodysplasia of the spinal cord and absence of neurological signs (the meningeal sac may or may not contain parts of the cord or nerve roots but if it does, they conduct impulses normally), and (b) myelomeningocele, in which there is lack of fusion of the vertebral arches with cystic distension of the meninges, myelodysplasia of the spinal cord, and neurological signs.

The finding of a pure myelomeningocele with a cystic cavity of meninges only, without involvement of the spinal cord or spinal roots, is relatively uncommon. In a study of 306 patients with SBC, Smith[1] could only classify 26 (8.5 percent) as having meningoceles on the basis of operative findings and normal neurological clinical state.

### *Incidence*

Smith[1] reported an incidence of SBC in 1.24 per 1,000 live births, or approximately 1 per 800 live births. It was estimated that about one-third of affected children would reach school age each year; two-thirds of these would have severe neurological disability, and the remainder would have minimal or no disability. With the declining incidence of poliomyelitis, myelomeningocele is now second in importance to cerebral palsy as a cause of chronic locomotor disability in childhood. There is a slightly higher incidence of myelomeningocele in females; out of 371 patients (consecutive cases) reported by Menelaus,[2] 56 percent were girls.

### *Causative Factors*

In a study of 192 families to whom a child with SBC was born, Smith[1] was unable to determine any exogenous factors which could be considered responsible for the development of the defect. He pointed out that in order to affect the spine, any influence on the developing embryo would have to occur in the first few weeks of pregnancy. The neural plate appears first in the late presomite phase, at about 18 days after conception, and the neural tube is completed just after somite formation ceases by the end of the fourth week.

The parents in his study appeared to belong to all levels of society, and no economic or environmental influence could be determined. There was no significant relationship between the development of SBC to birth order or maternal age at the time of birth. Although there was no relationship of the disorder to hydramnios, Smith cited reports in which there was a high incidence of congenital anomalies of the nervous system in hydramniotic pregnancies.

In 15 of 192 families (7.8 percent), there was more than one child in the same family born with SBC. In five other families (2.8 percent), there was a family history of the condition in a close relative.

In a study of families with central nervous system anomalies, Record and McKeown[3] found that 2.8 percent of siblings born after the first malformed child carried a nervous system defect. If no hereditary factor existed, the expected incidence of SBC was 0.45 percent, so there was approximately a six-fold risk compared with that in a normal family; the actual expectancy for a defective family was about 3 percent. The evidence from these and other studies indicates that the risk of having a second affected sibling is fairly high.

In addition to the risk of SBC in subsequent pregnancies, there is a high risk of other congenital anomalies (such as cleft palate, congenital heart disease, or mongolism). In the 192 families reviewed by Smith,[1] in addition to the 20 families with a familial history of SBC, another 15 families had a history of other congenital anomalies.

### *Clinical Features*

Because of the many systems involved, few diseases in medicine require the multiprofessional approach more than SBC. There is not only the defect of the spine and the associated mass on the

TABLE 1.—*Gradings of Disability Related to Spinal Segments in Myelomeningocele Patients (Smith[1])*

| Group | Level of Spinal Lesion | Clinical State |
|---|---|---|
| I | Above $L_3$ ..... | Total paraplegia |
| II | $L_4$ and below ... | Paralysis of hip extensors and knee flexors. Flail feet. Bladder and rectal incontinence. *But* retention of use of knee extensors and hip flexors and adductors. |
| III | $S_1$ and below ... | Weakness of hip extensors and knee flexors. Paralysis of plantar flexors of feet. Weak inversion and eversion of feet. Inability to spread toes or cup soles of feet. Bladder and rectal incontinence. *But* retention of use of knee extensors, hip flexors and adductors, and dorsiflexors of feet; and some use of hip extensors and knee flexors. |
| IV | $S_3$ and below ... | Bladder and rectal incontinence. *But* normality of lower limbs. |

back, but a high percentage has associated hydrocephalus and also neuromuscular, sensory, and autonomic disturbances, orthopedic problems, bowel and bladder incontinence, and often mental and emotional disturbance. The lumbar and lumbosacral regions of the spine are predominantly affected; 90 percent of the lesions occur below the second lumbar vertebra and 42 percent at the lumbosacral junction.

The muscles of the legs receive their innervation from more than one nerve root and approximately half of the nerve supply to an individual muscle must be lost before paralysis can be detected clinically. Most myelomeningoceles occur in the lumbosacral region and are usually associated with a lower motor neuron paralysis of leg muscles. Depending on the level of the defect, paralysis of muscles may be either flaccid or spastic. Sometimes there are contractures which produce flexion at the hip and extension at the knee. A variety of clubfoot deformities occur. Ankle jerks are usually absent but knee jerks are often present and active.

In a study of 295 patients, Menelaus[2] noted that gross neurological abnormalities were less common when the lesion was high in the spine. Of the three cervical lesions in his group of cases, only one was associated with abnormal neurological signs, and these were minimal. On the other hand, most (approximately 85 percent) of the lumbar and lumbosacral lesions were associated with extensive paralysis. Of 146 consecutive patients with myelomeningocele, 4 percent had minimal or no paralysis in the legs, 20 percent had no innervation below the twelfth thoracic nerve roots, 23 percent had none below the second lumbar nerve roots, and 53 percent had none below the second sacral nerve roots.

There was also a considerably higher mortality in patients with higher lesions—20 of 29 patients with no innervation below the twelfth thoracic roots had died, whereas only 24 of 77 with no innervation below the second sacral nerve roots had died.

When spina bifida affects the upper cervical segment, cerebellar ataxia may be present as well as weakness and spasticity of the legs and arms. The sensory disturbances are confined to the upper cervical dermatomes. Smith[1] has suggested a grading scale of disabilities related to spinal segments of myelomeningocele patients. He divides them into four groups, to highlight four different gradings of severity ranging from total paraplegia to the child who requires no locomotor support at all (Table 1).

Analgesia and loss of awareness to heat may be found over the ankles and feet and, in many cases, this sensory loss extends up over the posterior surface of the legs and thighs to the buttocks and perineum. Tactile sensibility is less affected than pain sense.

Proprioceptive awareness is usually not affected except in cases with severe involvement. The distribution of sensory disturbances is usually of the segmental type.

Usually sphincter control is inadequate unless the symptoms are very mild; complete incontinence of both bladder and bowel occurs if the legs are paralyzed. When the lesion is confined mainly to the conus, loss of sphincter control may be the only evidence of myelodysplasia; the anus is relaxed and the rectum may prolapse.

Hydrocephalus occurred in 77 percent of the cases in Menelaus' series[2] and its incidence was related to the neurological level, there being a 90 percent incidence in higher lesions and 50 percent in the lower ones.

In a group of 134 children who were five years of age or older, reported by Lorber,[4] the average intelligence quotient (IQ) of 59 children who had either no hydrocephalus, or moderate hydrocephalus which required no treatment, was 87 (range 50 to 129); 83 percent of the group had an IQ of

80 or more, and only 5 percent were severely retarded. Included in this study were another 75 children with surgically treated hydrocephalus whose IQ averaged 79 (range: too low to test, to 124); 53 percent had an IQ of 80 or more and 47 percent were retarded, including 17 percent who were grossly retarded.

*Selection of Cases for Treatment*

A child born with SBC faces a lifetime of being handicapped and being treated. Coping with life is difficult enough for those who are normal; how much more difficult when there is a motor disturbance, a sensory disorder, poor bowel and bladder control, and sometimes mental retardation! The family's coping ability is also severely tested when the child requires periodic surgical therapy, bracing, physical therapy, and special schooling, and may as an adult still continue to be dependent on others.

The survival rate of these children has increased in recent years, because more and more infants are being treated surgically. The medical attitude regarding treatment varies. Zachary[5] recommends that all affected patients be operated upon, even if many of the survivors will suffer from multiple handicaps. Matson[6] considers it wrong to treat infants whose predictable quality of life would be poor.

Lorber,[4] in referring to a personal communication from Ingram, indicates that there are several medical centers which exercise variable degrees of selection. A severe degree of paralysis is the most generally adopted reason for withholding treatment, but incontinence, severe hydrocephalus, gross deformities and poor social conditions are also taken into account. Lorber notes that some would prefer not to treat the most severe cases, but are afraid that surviving normal untreated infants would have greater handicaps than if they had been given total care. He emphasizes that the future quality of life of many of the patients with myelomeningocele depends, at least partly, on the speed, efficiency, and comprehensiveness of treatment from birth and often for the rest of their lives.

Obviously, many will still be severely physically handicapped or mentally retarded, or both, in spite of everything that is done. With the survival of these patients, there is a significant effect on the lives of both patient and family. The community also becomes involved in sharing in the cost of care and providing special programs, such as schools for the handicapped, out-of-home placement, and workshops and agencies which will provide appropriate emotional support through their social and guidance workers.

Lorber[4] made an attempt to determine whether it would be possible to foretell from simple physical signs on the first day of life what the likely future of a baby would be if he were untreated, and to compare this with his chances if he were given total care as it is known today. He analyzed two groups, one consisting of 323 infants treated between 1959 and 1963, and the other, of 201 infants treated during 1967-1968. In all cases, treatment was started on the first day of life, but the treatment for the first group was not considered to be as good as it was for the second group. In the first series, 50 percent survived to two years, compared with 64 percent in the second. The proportion with serious intellectual or physical handicaps did not decrease in the second series. Laurence[7] reported on the survival of untreated SBC in South Wales during a seven-year period from 1956 to 1961; out of 426 children born alive, only 25 percent were still living at two years of age.

Lorber[4] stated that a higher survival rate and a better quality of life would be possible with improved techniques and better application of existing knowledge and facilities. Some patients are so severely handicapped at birth that no method of therapy would prevent major permanent multiple handicaps, a short life, or a severely restricted quality of life. If we wished to spare children and their families prolonged suffering and to give better attention to those who are more likely to benefit from total care, Lorber[4] states that it may be necessary to select suitable cases for intensive treatment and to withold treatment from others. He felt that it was possible to forecast with accuracy, from purely clinical assessment, the minimal degree of future handicap in an individual, even if it is impossible to forecast the maximal degree of disability which he may suffer if he survives. He made clear that therapeutic efforts should be concentrated on those who could truly benefit from treatment, that is, the less severely affected. Lorber concluded his work by quoting Matson:[6]

"In our clinic, it is not customary to operate upon newborn infants or those in the first few months of life with thoraco-lumbar or upper-lumbar myelomeningoceles or myeloschises who exhibit complete sphincter paralysis and total

paraplegia below the upper lumbar segments. This is true whether or not there is significant hydrocephalus at the time. When examination in the first day of life, therefore, confirms total absence of neurologic function below the upper-lumbar levels, custodial care only is recommended. It is recognized that some of these totally paralyzed hydrocephalic patients recommended for custodial care survive for considerable periods of time. For these patients and their families, it is the doctor's and the community's responsibility to provide this care and to minimize suffering; but, at the same time, it is also their responsibility not to prolong such individual, familial, and community suffering unnecessarily, and not to carry out multiple procedures and prolonged, expensive, acute hospitalization in an infant whose chance for acceptable growth and development is negligible."

*Ethical Considerations*

If the decision is made not to treat, then what? Does the physician withhold parenteral fluids if the child does not suck well? Are antibiotics not to be prescribed if a treatable infection develops? Should palliative procedures be withheld—for example, repairing the skin defect on the back and shunting an enlarging head, even if more extensive therapy is not contemplated? If the child survives the first year, should he be refused other treatment—orthopedic, urologic, or whatever might make life more meaningful and more comfortable?

And what of the parents? If it is decided to do nothing, will they receive the same emotional support from the social workers and other members of the team as the family of the child who is undergoing aggressive therapy?

Who is to pay for the care of the child who is left to "sink or swim" in the family whose financial resources are limited and unlikely to be adequate for the costly nursing care that may be necessary? Will the community accept the financial responsibility to ensure that, if the child survives, life will be kept meaningful to him as an individual regardless of his intellectual limitations and physical handicaps?

There is no simple answer to these questions. My feeling is that these children and their families should be evaluated by a team, including at least a pediatrician, an orthopedist, a urologist, a neurosurgeon, and a social worker. After a careful appraisal of all factors, a tentative prognosis should be made; then one of the team members, acting as coordinator, should interpret the findings and prognosis to the parents while the others should be available for whatever specific help they can provide for the parents. The social worker should be involved in the discussions to be aware of what the parents are told; he can then interpret information, which may be confusing, and can continue to provide emotional support to them. On the basis of the prognosis and the outline of therapy that may be found to be necessary, the physician should assist the family in making a decision as to what treatment is to be carried out. Whatever decision the parents make, it is important that they be relieved of any sense of guilt.

Once the decision is made, the care of the child should proceed in the most humane way possible. Cost of the treatment should not be a limiting factor in the decision. Community support, such as that provided through Crippled Children's Services, should be available to all who need it. Help should not be limited to medical, surgical and hospital care, but should also include counseling and guidance by social workers and public health nurses, out-of-home nursing care whenever indicated, respite and homemaker services, and education and recreational services.

As long as modern ethical practice permits destruction through abortion of unborn children who are likely to be physically and mentally handicapped, and requires that liveborn, obviously defective children not be deliberately destroyed, then every effort should be made to make the life of the individual as meaningful as possible under the specific circumstances. The child should have as much freedom from pain and discomfort as medical and nursing care can provide, and the family should have an opportunity to share their burden of care with dignity and with compassionate assistance from the community.

**REFERENCES**

1. Smith ED: Spina Bifida and the Total Care of Spinal Myelomeningocele. Springfield, Ill, Charles C Thomas, 1965

2. Menelaus MB: The Orthopaedic Management of Spina Bifida Cystica, Edinburgh, E and S Livingstone, 1971

3. Record RG, McKeown T: Congenital malformations of the central nervous system—II. Maternal reproductive history and familial incidence. Br J Soc Med 4:26-50, 1950

4. Lorber J: Results of treatment of myelomeningocele—An analysis of 524 unselected cases, with special reference to possible selection for treatment. Dev Med Child Neurol 13:279-303, Jun 1971

5. Zachary RB: Ethical and social aspects of treatment of spina bifida. Lancet 2:274-276, Aug 3, 1968

6. Matson DD: Surgical treatment of myelomeningocele. Pediatrics 42:225-227, 1968

7. Laurence KM: The survival of untreated spina bifida cystica. Dev Med Child Neurol (Suppl 11):pp 10-19, 1966